

[CLAIMS]

[Claim 1] A method for diagnosing an inherited neuropathy, comprising:

running the PCR amplification using microsatellites present in the chromosome 17p11.2-p12 region as markers; and

5 DNA typing the resulting PCR amplification products to determine the presence of duplication and deletion in the corresponding chromosomal region,

wherein PCR amplification is carried out using 6 loci of D17S921, D17S9B, D17S9A, D17S918, D17S2230 and D17S4A as markers, and DNA-
10 typing of the resulting PCR amplification products is then carried out to determine duplication and deletion in the corresponding chromosomal region.

[Claim 2] The method according to claim 1, wherein PCR is carried out to simultaneously amplify 6 markers, using a mixture of primers of SEQ. ID.
15 NOS: 1 and 2; primers of SEQ. ID. NOS: 3 and 4; primers of SEQ. ID. NOS: 5 and 6; primers of SEQ. ID. NOS: 7 and 8; primers of SEQ. ID. NOS: 9 and 10; and primers of SEQ. ID. NOS: 11 and 12 in differential concentrations and a standard allele ladder.

20 [Claim 3] The method according to claim 1, wherein the method includes:

(a) PCR amplification of 3 markers and DNA typing of the resulting PCR amplification products, using a mixture of primers of SEQ. ID. NOS: 3 and 4; primers of SEQ. ID. NOS: 5 and 6; and primers of SEQ. ID. NOS: 7

and 8 in differential concentrations and a standard allele ladder, thereby firstly determining duplication and deletion in the 17p11.2-p12 region; and

(b) PCR amplification of the remaining 3 markers and DNA typing of the resulting PCR amplification products, using a mixture of primers of SEQ. ID. NOS: 1 and 2; primers of SEQ. ID. NOS: 9 and 10; and primers of SEQ. ID. NOS: 11 and 12 in differential concentrations and a standard allele ladder, thereby secondly determining duplication and deletion in the 17p11.2-p12 region.

10 [Claim 4] A kit for diagnosing an inherited neuropathy by determination of duplication and deletion in the chromosome 17p11.2-p12 region, comprising:

a mixture of primers of SEQ. ID. NOS: 1 and 2; primers of SEQ. ID. NOS: 3 and 4; primers of SEQ. ID. NOS: 5 and 6; primers of SEQ. ID. NOS: 7 and 8; primers of SEQ. ID. NOS: 9 and 10; and primers of SEQ. ID. NOS: 11 and 12 in differential concentrations; and
a standard allele ladder.

20 [Claim 5] A kit for diagnosing an inherited neuropathy by determination of duplication and deletion in a chromosome 17p11.2-p12 region, comprising:

a first kit including a mixture of primers of SEQ. ID. NOS: 3 and 4; primers of SEQ. ID. NOS: 5 and 6; and primers of SEQ. ID. NOS: 7 and 8 in differential concentrations and a standard allele ladder; and

a second kit including a mixture of primers of SEQ. ID. NOS: 1 and 2; primers of SEQ. ID. NOS: 9 and 10; and primers of SEQ. ID. NOS: 11 and 12 in differential concentrations and a standard allele ladder.